BIRTH DEFECT RISK FACTOR SERIES: CRANIOSYNOSTOSIS

DEFINITION

Craniosynostosis is premature closure of any of the cranial sutures (sagittal, coronal, lambdoidal, metopic). It can affect one suture or multiple sutures. Craniosynostosis is associated with several syndromes; the most commonly reported ones are Crouzon's disease, Apert syndrome (Acrocephalosyndactyly Type I), Carpenter syndrome (Acrocephalosyndactyly Type II), Saethre-Chotzen syndrome (Acrocephalosyndactyly Type III), and Pfeiffer syndrome (Acrocephalosyndactyly Type IV).

DEMOGRAPHIC AND REPRODUCTIVE FACTORS

Craniosynostosis risk appears to increase with increasing **maternal age** (Kallen, 1999; Singer, 1999; Alderman, 1988), and with increasing **paternal age** (Singer, 1999; Alderman, 1988).

One study reported a higher prevalence of craniosynostosis among non-blacks (Alderman, 1988). However, another investigation found no significant effect of **race/ethnicity** on craniosynostosis risk (Singer, 1999).

Sex influences craniosynostosis risk. Most studies have reported higher craniosynostosis rates among males, particularly for sagittal and lambdoidal craniosynostosis (Lary, 2001; Kallen, 1999; Singer, 1999; Alderman, 1988). However, the coronal craniosynostosis rate seems higher among females (Kallen, 1999; Lajeunie, 1995).

Parity does not appear to affect craniosynostosis prevalence (Kallen, 1999; Alderman, 1988). Evaluation of the impact of **plurality** on craniosynostosis risk has produced inconsistent results, with one study reported an increased risk for multiple births (Alderman, 1988) while others found no association (Singer, 1999; Kallen, 1986).

Place of residence has not been found to affect craniosynostosis risk (Singer, 1999; Alderman, 1988), except for an increased risk for coronal and lambdoidal craniosynostosis in urban areas (Singer, 1999). Also, an association between craniosynostosis and living at **high altitudes** has been reported (Alderman, 1988; Alderman, 1995), although one investigation failed to find such an association (Castilla, 1999).

One investigation reported a **secular trend**, with an increase in craniosynostosis rates over time. Most of the increase was due to an increase in the number of reported lambdoidal craniosynostosis cases (Singer, 1999).

FACTORS IN LIFESTYLE OR ENVIRONMENT

Neither **maternal** nor **paternal education** has been reported to affect craniosynostosis risk (Kallen, 1999; Alderman, 1988). **Maternal occupation** does not appear to influence craniosynostosis rates; however, increased rates for **paternal occupation** in the agriculture and forestry or mechanics and repairmen fields have been suggested (Bradley, 1995). One investigation reported no association between **parental farming** occupation and **pesticide** exposure and risk of craniosynostosis (Kristensen, 1997).

No relationship between maternal **hypothyroidism** or **hyperthyroidism** and craniosynostosis has been reported (Khoury, 1989).

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Several studies have found an association between craniosynostosis risk and maternal **smoking** (Honein, 2000; Kallen, 1999; Alderman, 1988; Alderman, 1995). However, no increased rates due to maternal **alcohol** consumption have been identified (Alderman, 1994). Maternal use of **nitrosatable drugs** has been linked to sagittal and lambdoidal craniosynostosis (Gardner, 1998).

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Please Note: *The primary purpose of this report is to provide background necessary for conducting cluster investigations. It summarizes literature about risk factors associated with this defect. The strengths and limitations of each reference were not critically examined prior to inclusion in this report. Consumers and professionals using this information are advised to consult the references given for more in-depth information.*

This report is for information purposes only and is not intended to diagnose, cure, mitigate, treat, or prevent disease or other conditions and is not intended to provide a determination or assessment of the state of health. Individuals affected by this condition should consult their physician and when appropriate, seek genetic counseling.

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